A Resource for Health Care Professionals
Screening and Diagnosis of Celiac Disease

A Summary from the NASPGHAN, WGO and ACG Guidelines

Canadian Celiac Association Professional Advisory Council, 25 May 2016

Celiac disease (CD) is an immune mediated injury to the small intestine that is caused by ingestion of gluten (a name for multiple proteins in wheat, rye and barley) in genetically susceptible individuals. In Canada, CD occurs in approximately 1:100 people. The disease can be difficult to diagnose because it presents with a variety of symptoms (see Table) at any point in life. Prevalence of CD is higher in specific associated conditions (see Table).

Practice Point: The classic form of CD can manifest at any age once foods containing wheat, barley or rye are introduced in the diet. Symptoms include weight loss, diarrhea, abdominal pain/distension, and occasionally, severe malnutrition. Most patients display one or more intestinal symptom(s), others present with non-gastrointestinal symptoms (see Table) and some may be overweight. Patients may have an associated condition with or without celiac-related symptoms (see Table). Iron or folate deficiency anemia can occur due to malabsorption. Children may present with short stature, delayed puberty or dental enamel defects. Many symptoms (for example, anemia, weight loss, bone pain, paresthesia, edema, skin changes) are secondary to nutrient deficiency states.

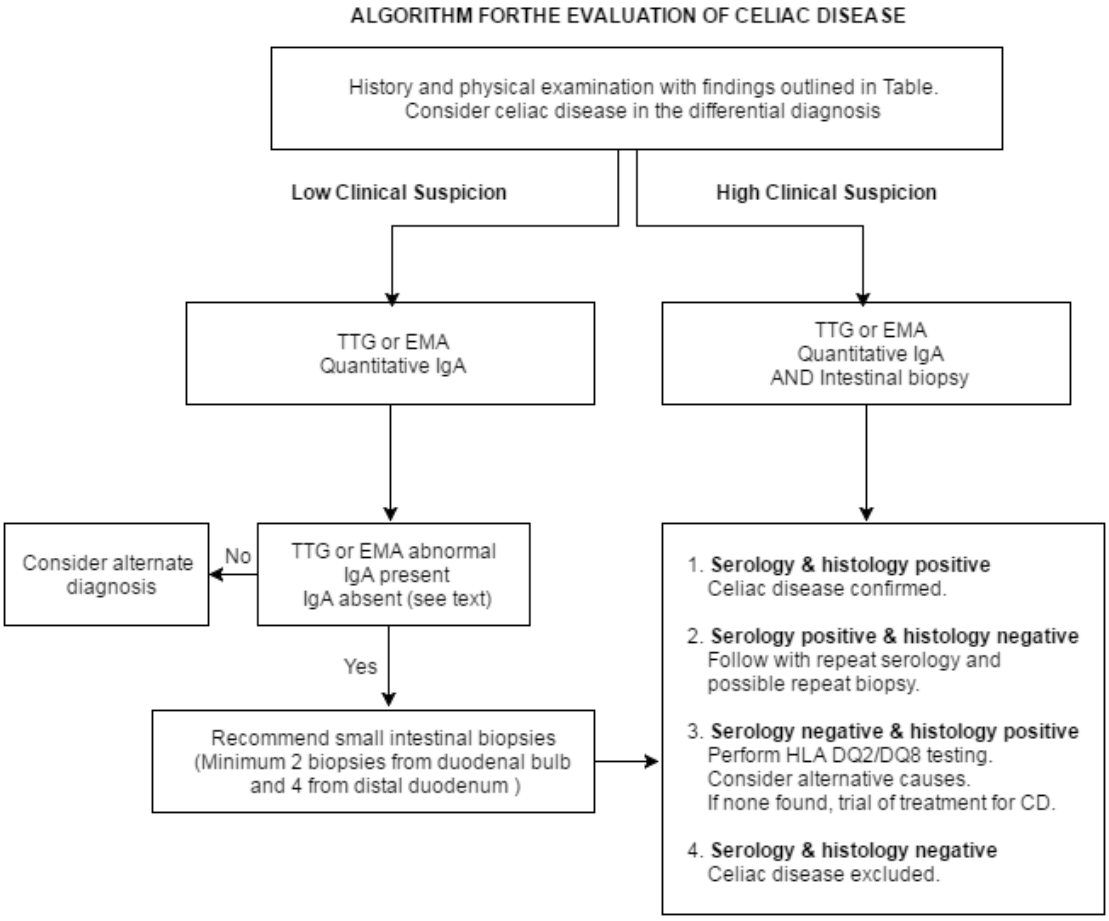
Classic Symptoms	**Non-classic Symptoms and Signs**
Abdominal distension Abdominal pain Chronic diarrhea Anorexia Irritability Weight loss (or failure to thrive in children) Muscle wasting Dermatitis herpetiformis (DH)	Unexplained iron or folate deficiency anemia Aphthous stomatitis (oral canker sores) Dental enamel defects Persistent/recurrent vomiting Irritable bowel syndrome Chronic constipation Abnormal liver enzymes (ALT/AST) Arthritis, arthralgia Osteoporosis/Osteopenia Short stature Delayed puberty Infertility
Associated Conditions (% affected)	**Neurological presentations**
Relative of individual with CD (8-15%) Type 1 diabetes mellitus (4-8%) Autoimmune thyroiditis (2-5%) Trisomy-21 (Down syndrome) (2-5%) Turner syndrome (2-5%) IgA deficiency (2-5%, up to 30% in patients with gastrointestinal symptoms)	Unexplained ataxia Peripheral neuropathy Epilepsy with occipital calcifications Depression, anxiety Fatigue

CD should be suspected in individuals with ANY of the above symptoms, signs or associated conditions. Screening serologic tests can be utilized to identify individuals at risk for CD. The diagnosis MUST be confirmed with a small intestinal biopsy or a skin biopsy in patients with DH. It is strongly recommended that the biopsy be done BEFORE starting on a gluten-free diet (GFD) because eliminating gluten can interfere with making an accurate diagnosis. CD requires lifelong treatment with a GFD. The diet is complicated, expensive and there are concerns about the nutritional adequacy of GF products as they can be high in fat and sugar, and often low in fiber, iron and B vitamins. Patients should be referred to a registered dietitian with expertise in CD and the GFD.

Screening tests and intestinal biopsy need to be performed while the patient is on a gluten-containing diet. Individuals who have started a GFD will require a gluten challenge to confirm the diagnosis. See the NASPGHAN or American College of

Gastroenterology Celiac Disease Guidelines referenced below for information on the gluten challenge that can take from three weeks to two years to reinjure the intestine.

The IgA tissue transglutaminase antibody (IgA-TTG) or endomysium antibody (IgA-EMA) tests are recommended for initial testing performed by experienced laboratories. The choice of test depends on availability and laboratory preference. Both tests display positive predictive values that vary from 15% (< 3 X upper limit of normal, ULN) to greater than 95% (>10 X ULN), depending on the antibody level. Since these tests are IgA based, they will be falsely negative in patients with IgA deficiency. The prevalence of IgA deficiency is higher in individuals with CD; therefore, screening for selective IgA deficiency should be performed at the same time as the serology tests (see note below). The IgA and IgG anti-gliadin antibody (AGA) tests are no longer recommended as screening tests for CD because of poor predictive values.



Note: CD occurs in 2-5% of people with selective IgA deficiency. All symptomatic IgA deficient patients should be referred for endoscopic small intestinal biopsies regardless of their serology results, as false negatives can occur. In asymptomatic individuals with IgA deficiency, the laboratory may be able to perform IgG-TTG or an IgG-deamidated gliadin peptide (IgG-DGP). Negative HLA-DQ2 or DQ8 genetic tests are helpful to exclude the diagnosis of CD because over 99% of patients with CD are positive for HLA-DQ2 or DQ8. However, approximately 30% of the general population tests positive for one of these HLA types and most do not develop CD. For management information, see Follow Up Management of Celiac Disease.

Primary author: Dr. J. Decker Butzner
Chair, CCA Professional Advisory Council

References

Hill ID, Dirks MH, Liptak GS, et al; Guideline for the diagnosis and treatment of celiac disease in children: recommendations of the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. J Pediatr Gastroenterol Nutr. 2005; 40(1):1-19. http://www.naspghan.org/content/51/en/Celiac-Disease
Bai. JC, Fried GR, et al; WGO-OMGE practice guideline: Celiac disease February 2005 http://www.worldgastroenterology.org/guidelines/global-guidelines/celiac-disease/celiac-disease-english.
Rubio-Tapia A, Hill ID, Kelly CP, Calderwood AH, Murray JA. American College of Gastroenterology. ACG clinical guidelines: diagnosis and management of celiac disease. Am J Gastroenterol. 2013; 108 (5):656-76. http://www.gi.org/guideline/diagnosis-and-management-of-celiac-disease/.
Rashid M, Lee J: Serologic testing in celiac disease: Practical guide for clinicians. Can Fam Physician January 2016 62: 38-43. URL: http://www.cfp.ca/content/62/1/38.full Accessed 06, Feb 2016.